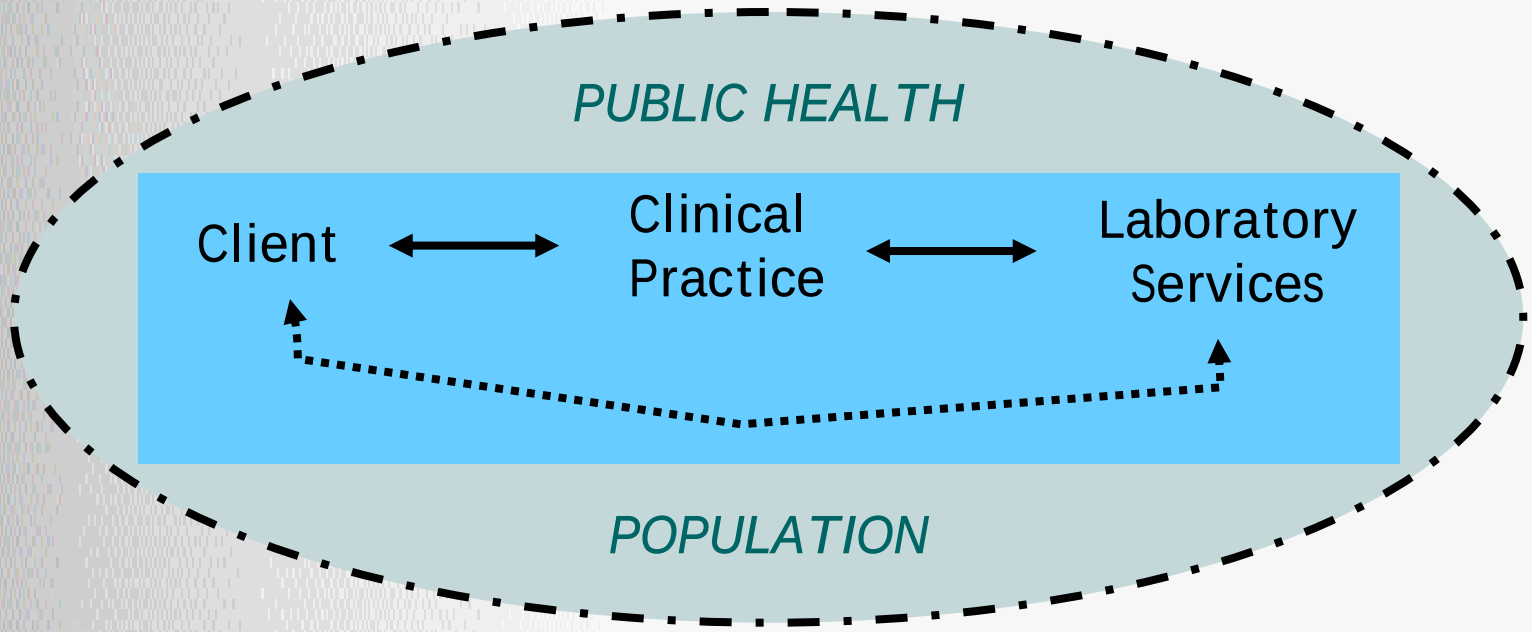
COMMUNICATION:
Key to Appropriate Genetic Test Referral, Result Reporting, Interpretation, and Use



Ira M. Lubin, PhD, FACMG
Laboratory Practice Evaluation and Genomics Branch
Division of Laboratory Systems
Public Health Practice Program Office
Centers for Disease Control and Prevention

U.S. Department of Health and Human Services

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Questions

How can we ensure that health-related decision making in clinical and laboratory practice is based upon the proper ordering, reporting, and use of genetic tests and results?

1. What variability exists in the ordering and reporting of genetic tests and results?

2. What issues have arisen within the clinical practice and laboratory setting in the use of genetic testing services?
   - patient outcomes
   - other costs

3. What efforts are can be considered to assure the appropriate ordering of genetic tests and reporting of results?
   - process issues
   - an informed/educated workforce and public

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Assessing Laboratory Practices

Quality Assurance in Molecular Genetic Testing Laboratories

Margaret M. McGovern, MD, PhD
Marta O. Benach
Sylvan Wallenstein, PhD
Robert J. Desnick, PhD, MD
Richard Keenlyside, MD, MS

JAMA. 1999;281:835-840

Context Specific regulation of laboratories ... be needed to ensure standards and quality assurance (QA) and safeguard patient rights to informed consent and confidentiality. However, comprehensive analysis of current practices of such laboratories, important for assessing the need for regulation and its impact on access to testing, has not been conducted.

Objective To collect and analyze data regarding availability of clinical molecular genetic testing, including personnel standards and laboratory practices.

(1999)

Personnel Standards and Quality Assurance Practices of Biochemical Genetic Testing Laboratories in the United States

Margaret M. McGovern, MD, PhD; Marta Benach, BA; Sylvan Wallenstein, PhD; Joe Boone, PhD; Ira M. Lubin, PhD

Arch Pathol Lab Med—Vol 127, January 2003

(2002)

article

September/October 2002 · Vol. 4 · No. 5

Medical genetic test reporting for cystic fibrosis (ΔF508) and factor V Leiden in North American laboratories

Hans C. Andersson, MD[1], Marie A. Krousel-Wood, MD, MSPH[2], Kelly E. Jackson, MS[1], Janet Rice, PhD[3], and Ira M. Lubin, PhD[4]

(2002)

article

May/June 2003 · Vol. 5 · No. 3

Physicians' perceived usefulness of and satisfaction with test reports for cystic fibrosis (ΔF508) and factor V Leiden

Marie Krousel-Wood, MD, MSPH[1], Hans C. Andersson, MD[2], Janet Rice, PhD[3], Kelly E. Jackson, MS[2], Eunice R. Rosner, EdD[4], and Ira M. Lubin, PhD[4]

(2003)

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Content Summary of Requisition Forms and Result Reports for Cystic Fibrosis Molecular Genetic Testing

Requisitions (N=17) (unpublished data (2003))	*Percent (N)*	Reports (N=28) (Genet Med (2002) 4:324)	*Percent (N)*
Indications for testing	88% (15)	Clinical Indications	64% (18)
Clinical information	59% (10)	Detection rate	86% (24)
Family information	41% (7)	Adjusted risk	71% (20)
Ethnicity	94% (16)	Ethnicity	21% (6)
Pedigree	47% (8)	Interpretation	93% (26)
Pregnancy status	59% (19)	Genetic Counseling	61% (17)

Follow-up study:

Physicians' Perceived Usefulness and Satisfaction with Test Reports for Cystic Fibrosis (ΔF508) and Factor V Leiden

In this study, we found physicians desired a more comprehensive report useful for guiding clinical decision-making.

(Genet Med (2003) 5:166)

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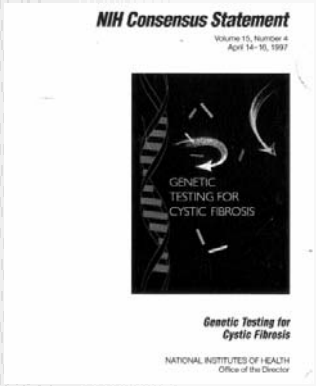
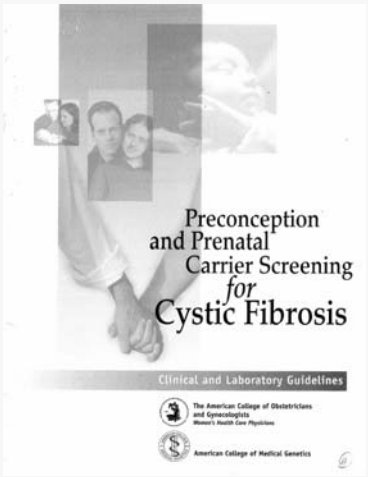
Some Recommendations / Standards Out there

CLIA

NCCLS

(1997)





(2001)

Vastag B (2003) Cystic fibrosis gene testing: experts say widespread use is creating unnecessary risks. *JAMA* 289:2923

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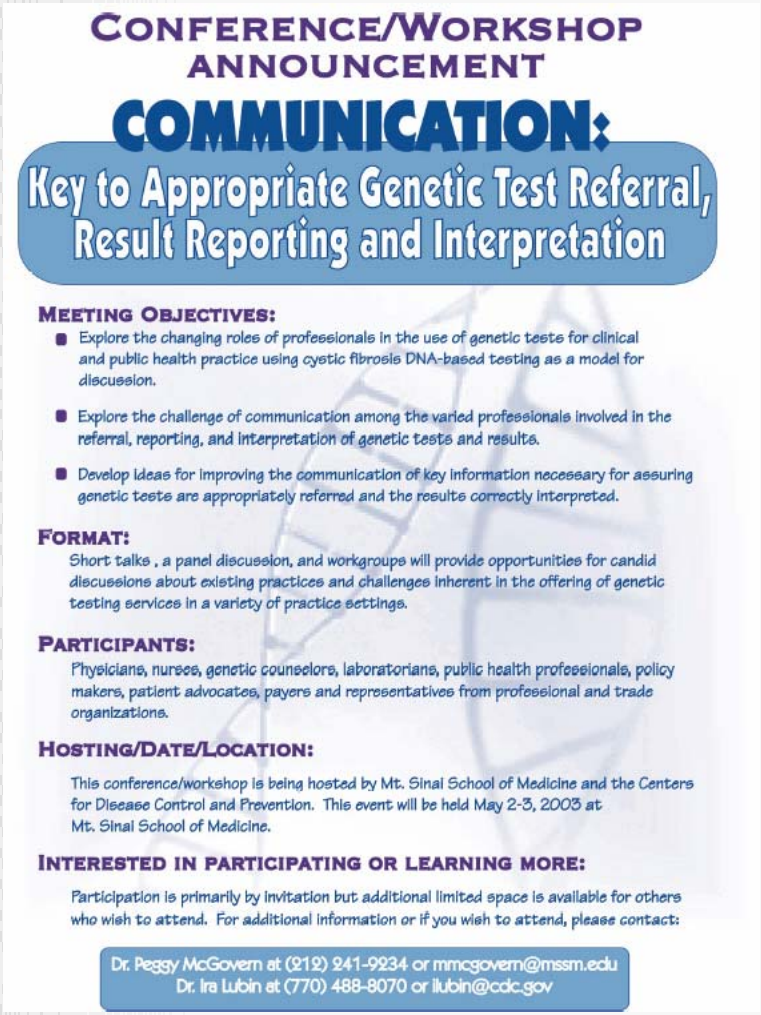
How can we ensure that health-related decision making in clinical and laboratory practice is based upon the proper ordering, reporting and use of genetic tests and results?



CONFERENCE/WORKSHOP ANNOUNCEMENT

COMMUNICATION:
Key to Appropriate Genetic Test Referral, Result Reporting and Interpretation

MEETING OBJECTIVES:

- Explore the changing roles of professionals in the use of genetic tests for clinical and public health practice using cystic fibrosis DNA-based testing as a model for discussion.
- Explore the challenge of communication among the varied professionals involved in the referral, reporting, and interpretation of genetic tests and results.
- Develop ideas for improving the communication of key information necessary for assuring genetic tests are appropriately referred and the results correctly interpreted.

FORMAT:

Short talks , a panel discussion, and workgroups will provide opportunities for candid discussions about existing practices and challenges inherent in the offering of genetic testing services in a variety of practice settings.

PARTICIPANTS:

Physicians, nurses, genetic counselors, laboratorians, public health professionals, policy makers, patient advocates, payers and representatives from professional and trade organizations.

HOSTING/DATE/LOCATION:

This conference/workshop is being hosted by Mt. Sinai School of Medicine and the Centers for Disease Control and Prevention. This event will be held May 2-3, 2003 at Mt. Sinai School of Medicine.

INTERESTED IN PARTICIPATING OR LEARNING MORE:

Participation is primarily by invitation but additional limited space is available for others who wish to attend. For additional information or if you wish to attend, please contact:

Dr. Peggy McGovern at (212) 241-9234 or mmcgovern@mssm.edu
Dr. Ira Lubin at (770) 488-8070 or ilubin@cdc.gov

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Conference Process

1. **Multi-disciplinary**

2. **One and a half day conference/workshop.**

3. **Orientation talks / panel discussion**

3. **Case-based discussion:**
    1. Carrier testing for cystic fibrosis with a relative with CBAVD
    2. Carrier testing for cystic fibrosis (without a family history for CF)
    3. Carrier testing for cystic fibrosis (with a family history)
    4. Diagnostic testing for cystic fibrosis - infant with failure to thrive
    5. Prenatal diagnosis

4. **Focus on pre- and post-analytic testing processes.**

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Organizational affiliation of Attendees

(attendance at the meeting does not imply endorsement)

Federal Government Agencies

Centers for Disease Control and Prevention, Department of Health and Human Services
Centers for Medicare and Medicaid Services, Department of Health and Human Services
Health Resources Services Administration, Department of Health and Human Services
Office of Science and Data Policy, Office of the Assistant Secretary for Planning and Evaluation
Department of Health and Human Services

Professional Organizations, Academics, and State entities

American Academy of Family Physicians
American Academy of Physician Assistants
American Association of Pediatrics
American College of Gynecology and Obstetricians
American College of Medical Genetics
American College of Nurse Midwives
American Medical Association
Association of Molecular Pathologists
Association of Public Health Laboratories
Association of Family Practice Residency Directors
Association of Women's Health, Obstetric, and Neonatal Nursing
Blue Cross and Blue Shield Association
GeneTests
Genetic Alliance
Genetics and Public Policy Center, Johns Hopkins University
International Society of Nurses in Genetics
March of Dimes Foundation
Minnesota Department of Health - Minnesota Children with Special Health Needs
Mount Sinai School of Medicine
National Coalition for Health Professional Education in Genetics
National Society of Genetic Counselors
New England Newborn Screening Program
St. Vincent's Hospital Cystic Fibrosis Center
Tulane University Health Sciences Center
Wadsworth Center, New York State Department of Health

International participation

Cystic Fibrosis Thematic Network
Organization for Economic Cooperation and Development

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A Few Major Issues and Recommendations

Issue: No standard format/process for requisitions and reports
Recommendation: Develop/evaluate standard practices

Issue: Limited data is available to quantify impact on patient outcomes and other costs.
Recommendation: Studies to collect and evaluate such data.

Issue: Practice/setting-specific guideline implementation and evaluation plans are virtually non-existent.
Recommendation: Develop such plans

Issue: Reimbursement issues
Recommendation: Provider/laboratory/payer forum

Issue: Role of the genetic laboratory - "Consultant" and/or "Provider" of test results
Recommendation: Develop provider/Laboratory partnerships

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A Few Words on Roles

1. **Physicians, physician assistants, nurses, other allied health professionals** have contact with patients.

2. **Geneticists/specialists** (clinical, laboratory, counselors, etc.) are far fewer in number.
   (i.e. laboratory's role- "consultant" and/or "provider" of test results)

3. **Public Health** provides assessment, policy, and assurance roles that can be critically important toward assuring the appropriate use of genetic testing.

4. **Consumers** are the decision makers

Next Steps

Domestic:

1. Document findings (conference summary)
2. Quantify impact of practices on patient outcomes and other costs
3. Development of appropriate standards/guidelines
4. Partnerships with organizations (follow up conference?)
5. Identify gaps in information being provided to the professionals and the general public for making educated and informed decisions.
6. Develop efforts to provide "missing information".
   - ⇒ community based
   - ⇒ make use of information technology tools
   - ⇒ evaluate usefulness

International:

1. Comparative international analysis of reporting practices
   (working with Cystic Fibrosis Thematic Network, Association of Molecular Pathologists, and Mt. Sinai School of Medicine)
2. Serving on the OECD* steering committee of quality assurance and proficiency schemes

(*Organization for Economic Cooperation and Development)

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International Efforts

MM1-A
Vol. 20 No. 7

Replaces MM1-P
Vol. 17 No. 21

Molecular Diagnostic Methods for Genetic Diseases; Approved Guideline

This document provides guidance for the use of molecular biological techniques for clinical detection of heritable mutations associated with genetic disease.

A guideline for global application developed through the NCCLS consensus process.

Clinical Molecular Genetics Society

Draft Best Practice Guidelines for Reporting

Payne S.
Kennedy-Galton Centre for Medical and Community Genetics, North West London Hospitals NHS Trust, London, United Kingdom.

These guidelines are an update of version 1 (issued January 1997:see CMGS website - www.cmgs.org) modified in light of experience scoring reports returned to the EQA steering committee during the 1996 and 1997 UK QA rounds. The aim has been to highlight some of the best (and worst) features of the reports returned. Guidelines prepared by Stewart Payne (s.j.payne@ic.ac.uk)..

http://www.emqn.org/Assets/uploadpdfs/REPORT.pdf

SGMG SSGM

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Merzenacker 8
CH 3006 Berne

Tél (41) 031 941 24 13 / Fax 941 24 15

Draft best practice guidelines on reporting in molecular genetic diagnostic laboratories in Switzerland

Document in preparation; this version last modified 23/11/2001.

http://www.ssgm.ch/sections/pdf/2001/meetings/GE01/DNA_reporting.pdf

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THANK YOU!

How can we ensure that health-related decision making in clinical and laboratory practice is based upon the proper ordering, reporting and use of genetic tests and results?

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